

**Table 2. SCA14 Genotypes and Phenotypes**

| Familial (F) / Sporadic (S) | Predicted Amino Acid Changes | Nucleotide Changes | Exon | Mean Age of Onset (Range in yrs) | Gait Ataxia | Dysarthria | Abnormal Eye Movement | Myoclonus       | Tremor/Chorea | Decreased Sense | Depression | Cognitive Deficits | Additional Signs          | Tendon Reflexes | MRI Cerebellar Atrophy | Author                                    |
|-----------------------------|------------------------------|--------------------|------|----------------------------------|-------------|------------|-----------------------|-----------------|---------------|-----------------|------------|--------------------|---------------------------|-----------------|------------------------|---|
| F                           | R41P                         | 122 G>C            | 1    | 46 (29-58)                       | +           | +          | -                     | -               | -             | -               | -          | -                  | -                         | nl              | +                      | Chen et al 2005                           |
| S                           | del100K -101H                | del296-301acaaac   | 4    | 38                               | +           | +          | +                     | -               | -             | -               | +          | -                  | -                         | ↑               | +                      | Chen et al 2005                           |
| F                           | H101Y                        | 301C>T             | 4    | 31 (10-50)                       | +           | +          | +                     | -               | -             | -               | -          | -                  | -                         | ↓/↑             | +                      | Chen et al 2003                           |
| F                           | H101Q                        | 303C>G             | 4    | 32 (40-20)                       | +           | +          | +                     | -               | -             | -               | -          | -                  | -                         | ?               | +                      | Alonso et al 2004                         |
| F                           | C114Y                        | 341G>A             | 4    | 28.8(17-37)                      | +           | +          | 2/5                   | -               | 2/5           | -               | -          | 1/5                | Extensor plantar reflexes | nl/↑            | +                      | Klebe et al 2005                          |
| F                           | G118D                        | 353G>A             | 4    | 38.3 (21-59)                     | +           | +          | +                     | 1/22            | -             | 8/22            | -          | -                  | Dystonia                  | nl/↑/↓          | +                      | Warrenburg et al 2003, Verbeek et al 2005 |
| F                           | S119P                        | 355T>C             | 4    | 42 (35-51)                       | +           | +          | -                     | -               | -             | +               | +          | -                  |                           | ↑               | +                      | Chen et al 2003                           |
| F                           | S119F                        | 356C>T             | 4    | 52.7 (14-70)                     | +           | -          | -                     | -               | -             | -               | -          | 1/5                | Seizures                  | nl/↑            | +                      | Hiramoto et al 2006                       |
| F                           | G123R                        | 367G>A             | 4    | 48.3 (40-60)                     | +           | +/-        | 2/3                   | -               | -             | 2/3             | -          | 1/3                | Deafness                  | ?               | +                      | Klebe et al 2005                          |
| F                           | G123E                        | 368G>A             | 4    | 36 (24-48)                       | +           | +/-        | -                     | 1/2             | 1/2           | 1/2             | -          | 2/2                |                           | ?               | +                      | Klebe et al 2005                          |
| F                           | Q127R                        | 380A>G             | 4    | 28 (12-42)                       | +           | +          | +                     | 5/11            | -             | -               | -          | -                  |                           | ↓               | +                      | Yabe et al 2003                           |
| S                           | G128D                        | 383G>A             | 4    | 22, 44                           | +           | +          | +                     | -               | -             | -               | -          | -                  |                           | nl              | +                      | Chen et al 2003; Morita et al 2006        |
| F                           | C131R                        | 391T>C             | 4    | Childhood-50                     | +           | +          | -                     | -               | -             | -               | -          | -                  | Seizures?                 | ?               | +                      | Dalski et al 2006                         |
| F                           | V138E                        | 413T>A             | 5    | 22(3-45)                         | +           | +/-        | +                     | 1/7             | 2/7           | 2/7             | -          | -                  | Extensor plantar reflexes | nl/↑            | +                      | Vlak et al 2006                           |
| F                           | C150F                        | 449,450gc>tt       | 5    | 31.3 (19-44)                     | +           | +/-        | +                     | -               | 1/6           | 4/6             | -          | -                  |                           | ↑               | +                      | Fahey et al 2005                          |
| F                           | G360S                        | 1078G>A            | 10   | 36.5 (20-53)                     | +           | +/-        | -                     | -               | 2/2           | -               | -          | -                  | Rippling                  | ?               | +                      | Klebe et al 2005                          |
| F                           | S361G                        | 1081A>G            | 10   | 28 (5-60)                        | +           | +          | -                     | -               | -             | -               | +          | -                  |                           | nl              | +                      | Chen et al 2005                           |
| F                           | F643L                        | 1928T>C            | 18   | 34 (childhood-60)                | +           | +/-        | 10/18                 | Facial myokymia | 4/18          | 4/18            | -          | 13/18              |                           | ↑               | +                      | Stevanin et al 2004                       |
| F                           | F643L                        | 1928T>C            | 18   | 20                               | +           | +          | -                     | -               | -             | +               | -          | -                  |                           | ?               | +                      | Klebe et al 2005                          |
| F                           | V692G                        | 2075T>G            | 18   | 15                               | +           | +          | -                     | -               | 2/2           | -               | -          | NA                 | Extensor plantar reflexes | ↑               | +                      | Klebe et al 2005                          |

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